Question 9 – October 26

A 65 year old man went to a dermatology office for evaluation of abnormal skin pigmentation. He had noticed a mole on his left shoulder that developed irregular margins and increased in size over the last 6 months. A comprehensive dermatologic examination revealed more than 50 similar but smaller lesions on his entire body. His father and paternal uncle have a history of melanoma in their mid-50s. Genetic testing confirmed CDKN2A mutation. Apart from melanoma, this patient is at highest risk of developing which other cancer?

A. Prostate cancer
B. Esophageal cancer
C. Gastric cancer
D. Pancreatic cancer
E. Colorectal cancer

Answer: D

This patient meets the criteria (history of melanoma in first or second degree relative, high total body nevi count usually >50, and nevi with concerning histological features under microscopy) for the diagnosis of familial atypical multiple mole melanoma syndrome. This is an autosomal dominant disease with variable penetrance. CDKN2A mutations are characteristic of this syndrome. Apart from melanoma, these patients are at risk of developing pancreatic cancer.

Reference:

Eckerle Mize D, Bishop M, Resse E, et al. Familial Atypical Multiple Mole Melanoma Syndrome. In: Riegert-Johnson DL, Boardman LA, Hefferon T, et al., editors. Cancer Syndromes [Internet]. Bethesda (MD): National Center for Biotechnology Information (US); 2009-. Available from: http://www.ncbi.nlm.nih.gov/books/NBK7030/